RFXANK gene

regulatory factor X associated ankyrin containing protein

Normal Function

The *RFXANK* gene provides instructions for making a protein that primarily helps control the activity (transcription) of genes called major histocompatibility complex (MHC) class II genes. Transcription is the first step in the production of proteins, and RFXANK is critical for the production of specialized immune proteins called MHC class II proteins from these genes.

The RFXANK protein is part of a group of proteins called the regulatory factor X (RFX) complex. This complex attaches to a specific region of DNA involved in the regulation of MHC class II gene activity. RFXANK helps the complex attach to the correct region of DNA. The RFX complex attracts other necessary proteins to this region and helps turn on MHC class II gene transcription, allowing production of MHC class II proteins.

MHC class II proteins are found on the surface of several types of immune cells, including white blood cells (lymphocytes) that are involved in immune reactions. These proteins play an important role in the body's immune response to foreign invaders, such as bacteria, viruses, and fungi. To help the body recognize and fight infections, MHC class II proteins bind to fragments of proteins (peptides) from foreign invaders so that other specialized immune system cells can interact with them. When these immune system cells recognize the peptides as harmful, they trigger the lymphocytes and other immune cells to launch immune responses to get rid of the foreign invaders.

The RFX complex also appears to play a role in the transcription of MHC class I genes, which provide instructions for making immune system proteins called MHC class I proteins. Like MHC class II proteins, MHC class I proteins attach to peptides from foreign invaders and present them to specific immune system cells. These cells then attack the foreign invaders to rid them from the body. While the RFX complex is able to help control MHC class I gene activity, it is not the primary regulator of these genes. Other proteins play a more prominent role in their transcription.

Health Conditions Related to Genetic Changes

bare lymphocyte syndrome type II

Mutations in the *RFXANK* gene are the most common genetic cause of an immune system disorder known as bare lymphocyte syndrome type II (BLS II). More than 40 mutations in this gene have been identified in affected individuals. BLS II is a type of combined immunodeficiency (CID), in which affected individuals have virtually no immune protection from foreign invaders. Consequently, individuals with BLS II have

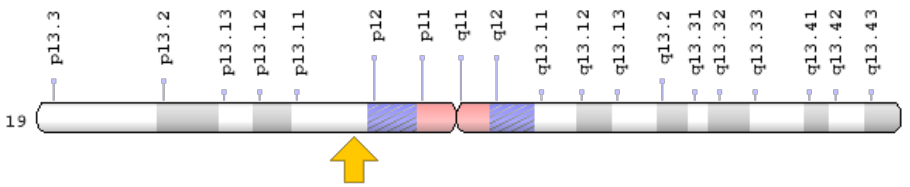
persistent infections in the respiratory, gastrointestinal, and urinary tracts, which can be life-threatening.

Mutations in the *RFXANK* gene lead to production of an altered protein that likely does not function properly. These changes impair binding of the RFX complex to DNA, which prevents transcription of MHC class II proteins. Consequently, lymphocytes lack any MHC class II proteins on their surface, and the body has difficulty getting rid of bacteria, viruses, and fungi, leading to the persistent infections characteristic of BLS II.

Chromosomal Location

Cytogenetic Location: 19p13.11, which is the short (p) arm of chromosome 19 at position 13.11

Molecular Location: base pairs 19,192,199 to 19,201,869 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ANKRA1
- ankyrin repeat-containing regulatory factor X-associated protein
- ankyrin repeat family A protein 1
- F14150_1
- MGC138628
- regulatory factor X subunit B
- RFX-B
- RFX-Bdelta4

Additional Information & Resources

Educational Resources

- Immunobiology: The Immune System in Health and Disease (fifth edition, 2001): The Major Histocompatibility Complex and Its Functions
  https://www.ncbi.nlm.nih.gov/books/NBK27156/
- Molecular Biology of the Cell (fourth edition, 2002): T Cells and MHC Proteins
  https://www.ncbi.nlm.nih.gov/books/NBK26926/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28RFXANK%5BTIAB%5D%29+OR+%28regulatory+factor+X+associated+ankyrin+containing+protein%5BTIAB%5D%29%29+OR+%28%28ANKRA1%5BTIAB%5D%29+OR+%28RFX-B%5BTIAB%5D%29+OR+%28ankyrin+repeat+family+A+protein+1%5BTIAB%5D%29+OR+%28ankyrin+repeat-containing+regulatory+factor+X-associated+protein%5BTIAB%5D%29+OR+%28regulatory+factor+X+subunit+B%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- REGULATORY FACTOR X, ANKYRIN REPEAT-CONTAINING
  http://omim.org/entry/603200

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_RFXANK.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RFXANK%5Bgene%5D
- HGNC Gene Family: Ankyrin repeat domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/403
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9987
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8625
- UniProt
  http://www.uniprot.org/uniprot/O14593

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